Function of cancer genes discovered

May 13 2005

Dutch researcher Sebastian Nijman has discovered new genes which are involved in the development of cancer. The results of his research have led to a new treatment for patients with an inherited form of cancer, cylindromatosis.
Patients with the very rare genetic condition cylindromatosis develop a lot of benign tumours on the skin. These tumours mainly occur on the head where they can cause serious malformations.

People with this disease have a mutated form of the protein CYLD. Nijman and his colleagues used genetic screens to discover the molecular mechanism underlying cylindromatosis. Nijman's research revealed that the CYLD protein plays an important role in the NF-kappa B signalling route. This is a cellular communications system which becomes overactive if the CYLD protein is mutated. This results in increased cell growth and the occurrence of tumours.

An important implication of this research is that inhibition of the NF-kappa B route in cylindromatosis patients could be an adequate form of treatment. Aspirin is a well-known inhibitor of this route and a clinical study into the effectiveness of aspirin ointment in the treatment of cylindromatosis is currently being carried out at the Netherlands Cancer Institute - Antoni van Leeuwenhoek Hospital.

Fanconi anaemia

The researcher also investigated another form of inherited cancer, Fanconi anaemia. People with this disease develop many highly

malignant tumours at a young age, because the cell repair system is incapble of detaching two linked DNA strands.

FANCD2 is a protein responsible for the repair of this DNA damage. The protein is linked to a second protein, ubiquitin, when the cell detects that DNA damage has occurred. Nijman identified a third protein, USP1, which detaches the FANCD2 from the ubiquitin. USP1 therefore plays a role in the repair of DNA damage and possibly in the development of cancer.

Sebastian Nijman's research was partially funded by NWO.

Source: NWO (Netherlands Organization for Scientific Research)